California Department of Health Services
Genetic Disease Branch
Expanded AFP Screening Program

850 Marina Bay Parkway, F175, Richmond, CA 94804 ● Phone 510/412-1502 ● Fax 510/412-1547

CALIFORNIA EXPANDED AFP SCREENING PROGRAM UPDATE

FEBRUARY, 2007

There are several new developments regarding prenatal screening in California:

1. NTD/SLOS Screening in Second Trimester

The Genetic Disease Branch is pleased to announce that, for specimens received as of February 10, 2007, the California Expanded AFP Screening Program will offer clinicians and their patients an "NTD/SLOS Screening" test in the second trimester. This was previously offered by the Program only to patients with Chorionic Villus Sampling (CVS) testing in the first trimester.

"NTD/SLOS Screening" is a blood test only for patients who have had First Trimester Screening, Chorionic Villus Sampling (CVS), or Preimplantation Genetic Diagnosis (PGD). Since women who have had any of these tests have already been screened or tested for chromosomal abnormalities, a woman and her clinician may now choose to have only NTD/SLOS screening in the second trimester.

The same AFP form currently being used for regular Expanded AFP Screening can also be used for "NTD/SLOS Screening". **If you mark question # 12 "YES"** on the AFP form (which is currently worded for CVS), your patient will receive risk assessments for:

***Open Neural Tube Defects**
*** SLOS (Smith-Lemli-Opitz Syndrome)**

If either result is "*Screen Positive*", the Expanded AFP Screening Program will pay for authorized follow-up services at a State-approved Prenatal Diagnosis Center.

If you mark question # 12 "NO" on the AFP form, the patient will receive Expanded AFP Screening (full triple-marker screening), i.e. the blood test results will include risk assessments for the above two conditions plus Down syndrome and Trisomy 18.

If you offer your patients the second trimester "NTD/SLOS Screening" option, please download a supply of *Patient Information and Consent/Refusal Form for NTD/SLOS Screening* from our web site www.dhs.ca.gov/gdb (Select "Prenatal Screening Section", then "Information for Patients"); or order them from our AFP Supplies desk phone 510-412-1441 or toll free 866-718-7915, select #3, or fax 510-412-1553. The Patient Information and Consent/Refusal Form for NTD/SLOS Screening is intended to be a supplement to the Expanded AFP Screening Program's Basic Booklet (patients under 35) or Choices Booklet (patients 35 or older). Patients must still receive the current booklets.

Summary: If a prenatal patient has previously had First Trimester Screening, CVS or PGD, and wishes to have NTD/SLOS Screening through the Expanded AFP Screening Program:

1. Provide the patient with the appropriate Expanded AFP "Basic Booklet" or "Choices Booklet".
2. Have the patient read the Patient Information and sign the Consent Form for NTD/SLOS Screening.
3. On the AFP Form, answer Question #12 "YES". If the form has wording for CVS only, you may write "First Trimester Screening" or "PGD" on the margin, if appropriate.
4. Draw the patient's blood between 15 - 20 weeks 0 days of pregnancy.

Please advise the staff who fill out the AFP form how to answer question # 12 so you and your patient will get the correct screening test! New AFP forms and patient booklets will be available in the near future.

(CONTINUED ON OTHER SIDE)

2. Note for hCG MoM >2.0 will no longer be included on the result mailer

For several years, the Expanded AFP Screening Program has noted when the hCG MoM (Multiple of the Median) is greater than 2.0 (single fetus), by adding an "Associated Complications" note at the bottom of the Expanded AFP result mailers. After data review and recommendations from the Program's consultant perinatologists, the Program has determined that this information is of limited use to prenatal care providers, particularly since there is no consensus on medical follow-up.

The actual hCG MoM is already on the result mailer, along with the hCG analytical value, allowing clinicians to determine for themselves whether the hCG MoM is high enough to warrant antepartum follow-up testing. Therefore, the note at the bottom of the result mailers (entitled "Associated Complications") which appears when the hCG MoM is greater than 2.0, will no longer be included for specimens received as of February 10, 2007.

3. Future Plans for Prenatal Screening

The California Expanded AFP Screening Program is planning to add a fourth marker, Inhibin, to the triple marker screening that now includes AFP, hCG, and uE3. This program expansion to second trimester Quad Marker screening is currently planned for July 2007.

There are also plans to expand the California Prenatal Screening Program even further the following year by offering first trimester screening in addition to second trimester screening. This will allow earlier risk assessment for chromosomal abnormalities through the California Program. This program expansion is currently planned for late summer 2008.

The Program will provide updates as these new developments are implemented.

For questions about any of these Program changes, please call The Expanded AFP Screening Program at 510-412-1502, or toll free 866-718-7915, press 5. Or call your AFP Coordinator, whose office phone number is listed at the bottom of every Expanded AFP result mailer.